Abstract

Inherited disorders of bilirubin metabolism – hereditary hyperbilirubinemias – are metabolic disorders manifested in early childhood. Unconjugated hyperbilirubinemias result from the defect of the enzyme uridine diphosphoglucuronosyltransferase (UGT1A1). UGT1A1 mediates the conjugation of bilirubin with glucuronid acid in hepatocytes and its elimination to water soluble compound. In the next step of bilirubin degradation the transport of conjugated bilirubin from hepatocyte into the bile occure. It is caused by the ATP dependent transporters ABCC2, ATP1B1 and OATP1B3. Mutations in the genes coding the bilirubin transporters results in conjugated hyperbilirubinemia Dubin-Johnson or Rotor syndrome. This study is focused on unconjugated hyperbilirubinemia in adolescents including the non-typical manifestations and the defects of ABCC2 transporter and their phenotype in humans.